

DFNB31 Antibody (Center) Blocking peptide

Synthetic peptide Catalog # BP11113c

Specification

DFNB31 Antibody (Center) Blocking peptide - Product Information

Primary Accession

Q9P202

DFNB31 Antibody (Center) Blocking peptide - Additional Information

Gene ID 25861

Other Names

Whirlin, Autosomal recessive deafness type 31 protein, DFNB31, KIAA1526, WHRN

Format

Peptides are lyophilized in a solid powder format. Peptides can be reconstituted in solution using the appropriate buffer as needed.

Storage

Maintain refrigerated at 2-8°C for up to 6 months. For long term storage store at -20°C.

Precautions

This product is for research use only. Not for use in diagnostic or therapeutic procedures.

DFNB31 Antibody (Center) Blocking peptide - Protein Information

Name WHRN (HGNC:16361)

Function

Involved in hearing and vision as member of the USH2 complex. Necessary for elongation and maintenance of inner and outer hair cell stereocilia in the organ of Corti in the inner ear. Involved in the maintenance of the hair bundle ankle region, which connects stereocilia in cochlear hair cells of the inner ear. In retina photoreceptors, required for the maintenance of periciliary membrane complex that seems to play a role in regulating intracellular protein transport.

Cellular Location

Cytoplasm {ECO:0000250|UniProtKB:Q80VW5}. Cell projection, stereocilium

{ECO:0000250|UniProtKB:Q80VW5}. Cell projection, growth cone

{ECO:0000250|UniProtKB:Q80VW5}. Photoreceptor inner segment

{ECO:0000250|UniProtKB:Q80VW5}. Synapse {ECO:0000250|UniProtKB:Q810W9}.

Note=Detected at the level of stereocilia in inner and outer hair cells of the cochlea and vestibule Localizes to both tip and ankle-link stereocilia regions. Colocalizes with the growing ends of actin filaments. Colocalizes with MPP1 in the retina, at the outer limiting membrane (OLM), outer plexifirm layer (OPL), basal bodies and at the connecting cilium (CC). In photoreceptors, localizes at a plasma membrane microdomain in the apical inner segment that surrounds the connecting cilia called periciliary membrane complex. {ECO:0000250|UniProtKB:Q80VW5, ECO:0000250|UniProtKB:Q810W9, ECO:0000269|PubMed:17584769}



DFNB31 Antibody (Center) Blocking peptide - Protocols

Provided below are standard protocols that you may find useful for product applications.

• Blocking Peptides

DFNB31 Antibody (Center) Blocking peptide - Images

DFNB31 Antibody (Center) Blocking peptide - Background

This gene is thought to function in the organization and stabilization of sterocilia elongation and actin cystoskeletalassembly, based on studies of the related mouse gene. Mutations in this gene have been associated with autosomal recessive non-syndromic deafness and Usher Syndrome. Alternative splicing of this gene results in multiple transcript variants encoding different isoforms.

DFNB31 Antibody (Center) Blocking peptide - References

Letra, A., et al. Am. J. Med. Genet. A 152A (7), 1701-1710 (2010) :Secolin, R., et al. Psychiatr. Genet. 20(3):126-129(2010)Aller, E., et al. Mol. Vis. 16, 495-500 (2010) :Toiyama, Y., et al. Int. J. Oncol. 35(4):709-715(2009)Gosens, I., et al. Hum. Mol. Genet. 16(16):1993-2003(2007)